COL9A1 gene

collagen type IX alpha 1 chain

Normal Function

The *COL9A1* gene provides instructions for making part of a large molecule called type IX collagen. Collagens are a family of proteins that strengthen and support connective tissues, such as skin, bone, cartilage, tendons, and ligaments. In particular, type IX collagen is an important component of cartilage, which is a tough, flexible tissue that makes up much of the skeleton during early development. Most cartilage is later converted to bone, except for the cartilage that continues to cover and protect the ends of bones and is present in the nose and external ears.

Type IX collagen is made up of three proteins that are produced from three distinct genes: one α1(IX) chain, which is produced from the *COL9A1* gene, one α2(IX) chain, which is produced from the *COL9A2* gene, and one α3(IX) chain, which is produced from the *COL9A3* gene. Type IX collagen is more flexible than other types of collagen molecules and is closely associated with type II collagen. Researchers believe that the flexible nature of type IX collagen allows it to act as a bridge that connects type II collagen with other cartilage components. Studies have shown that type IX collagen also interacts with the proteins produced from the *MATN3* and *COMP* genes.

Health Conditions Related to Genetic Changes

multiple epiphyseal dysplasia

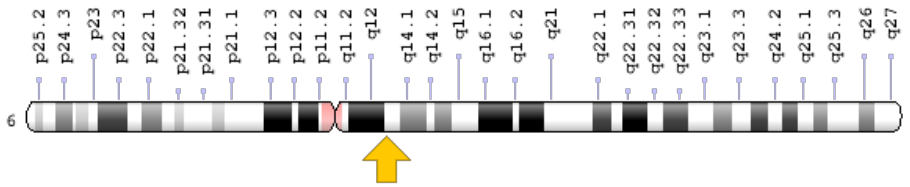
At least one mutation in the *COL9A1* gene has been found to cause dominant multiple epiphyseal dysplasia, a disorder of cartilage and bone development that primarily affects the ends of the long bones in the arms and legs (epiphyses). The identified mutation, called a splice-site mutation, involves the addition of one DNA building block (nucleotide) near an area of the gene called exon 8 (written as 1-bp ins, IVS8, T, +3). This mutation disrupts the way the gene's instructions are used to make the α1(IX) chain, resulting in a deletion of several protein building blocks (amino acids). It is not known how this mutation in *COL9A1* causes the signs and symptoms of dominant multiple epiphyseal dysplasia.

Stickler syndrome

Chromosomal Location

Cytogenetic Location: 6q13, which is the long (q) arm of chromosome 6 at position 13

Molecular Location: base pairs 70,216,040 to 70,303,083 on chromosome 6 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- alpha 1 type IX collagen
- cartilage-specific short collagen
- collagen IX, alpha-1 polypeptide
- collagen type IX alpha 1
- collagen, type IX, alpha 1
- DJ149L1.1.2
- FLJ40263
- MED

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Collagens Are the Major Proteins of the Extracellular Matrix
  https://www.ncbi.nlm.nih.gov/books/NBK26810/#A3551
- Molecular Cell Biology (fourth edition, 2000): Collagen: The Fibrous Proteins of the Matrix
  https://www.ncbi.nlm.nih.gov/books/NBK21582/

GeneReviews

- Multiple Epiphyseal Dysplasia, Dominant
  https://www.ncbi.nlm.nih.gov/books/NBK1123
- Stickler Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1302

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28COL9A1%5BTIAB%5D%29+OR+%28%28multiple+epiphyseal+dysplasia%5BTIAB%5D%29+OR+%28collagen+IX%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D

OMIM

- COLLAGEN, TYPE IX, ALPHA-1
  http://omim.org/entry/120210

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=COL9A1%5Bgene%5D
- HGNC Gene Family: Collagen proteoglycans
  http://www.genenames.org/cgi-bin/genefamilies/set/575
- HGNC Gene Family: Collagens
  http://www.genenames.org/cgi-bin/genefamilies/set/490
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2217
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/1297
- UniProt
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